bionano™

Bionano VIA™ Software Version 7.1 Release Notes

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Revision History

REVISION	NOTES
A	Initial release.

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Bionano VIA^{TM} Software

This document describes the v7.1 release of VIA software. This document provides an overview of what is changing with this release so that users may better understand the impact of moving to this version. Should users have any questions please contact software-support@bionano.com.

Introduction

Variant Intelligence Applications™ (VIA) software is a complete and integrated solution for the visualization, interpretation and reporting of genomic variants from multiple technology types. The predecessor version of VIA 7.1 is VIA 7.0. By supporting multiple genome-wide data modalities, VIA provides the most comprehensive view of genomic variants of any interpretation, annotation, and reporting software tool available. As a platform-agnostic tertiary analysis solution, VIA stores and manages distinct types of genomic data from various platforms enabling the extraction of meaningful insights from standalone or combined analysis.

The software includes algorithms to detect copy number variants (CNV) from major microarray vendors, optical genome mapping (OGM), and next generation sequencing (NGS) methodologies as well as Absence of Heterozygosity (AOH), from data types that assess B-allele frequency. VIA also provides interpretation assistance to analyze CNVs, Loss of Heterozygosity (LOH) and Structural Variants (SV) from OGM data. As a centralized analysis solution spanning technologies and application areas, VIA provides an efficient environment to keep pace with advancements in technology while retaining access to historical platform data. By being adaptive to whichever technology is used to generate CNV, LOH, or SV genomic variants, VIA provides rich annotations for the co-analysis of sequence variants from NGS to provide a complete picture of genomic variation and reveal more answers for disease association.

Supported Implementation Options

VIA is a scalable enterprise solution comprised of three components: the VIA Server, VIA Processing, and VIA Client applications. The server is the primary component of the system that manages the data in the repository. Installed adjacent to the Server is the Processing application that executes processing jobs for segmentation and annotation. The Client software is the User Interface (UI) utilized by all users to access sample data hosted in the VIA Server. There are three implementation options to deploy the VIA system; installation of the VIA Server and VIA Processing on the Bionano Access Server (BAS), local network server, or hosted in a cloud environment as illustrated below in **Figure 1**. In each implementation option, the VIA Client software is installed locally on each analyst's workstation with network access to the VIA Server. Further information on hardware requirements is available in the *Bionano VIA System Requirements* guides (CG-30577 and CG-30580).

- Option 1: VIA Server and VIA Processing are hosted on the BAS and is a common installation strategy for OGM users primarily interested in analyzing OGM data in VIA software.
- Option 2: VIA Server and VIA Processing are hosted on a local network and is a common installation strategy for NxClinical users primarily interested in array or NGS technology.
- Option 3: VIA Server and VIA Processing are hosted on a site-administered cloud environment and is a common installation strategy for users seeking production-scale analysis with VIA.
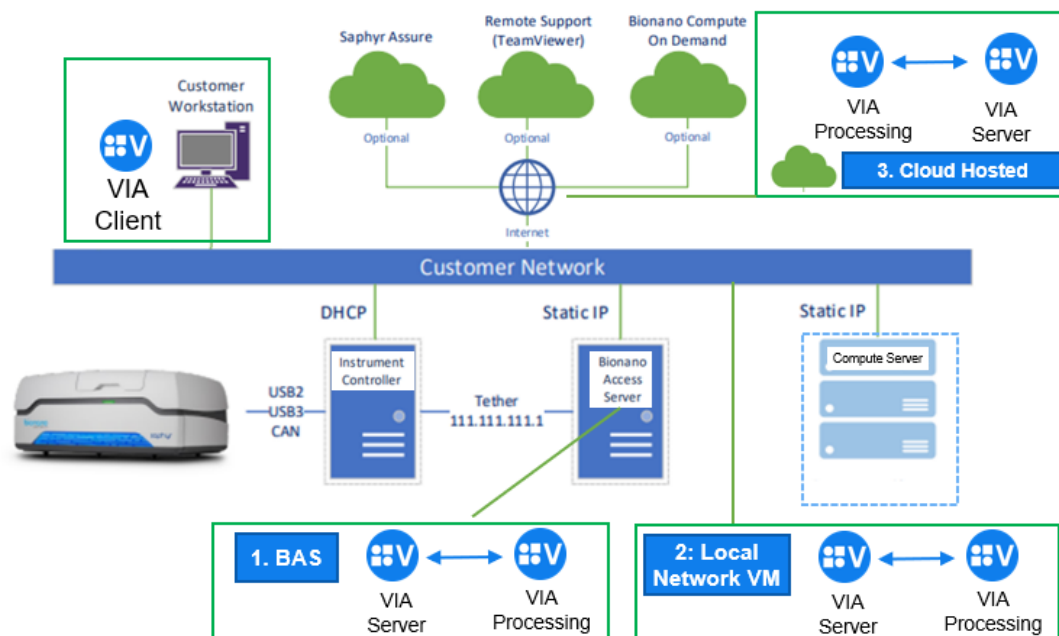
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Figure 1. VIA implementation options illustration

Compatibility

VIA 7.1 is compatible with Bionano Access™ 1.8+ and the Bionano Solve® v3.8+ pipeline running on either Saphyr® Compute, Bionano Compute servers or Bionano Compute On Demand v1.4. Please refer to the *Bionano VIA System Requirements* guide (CG-30577) for hardware requirements to install the software on local network and client workstations.

New Features in Version 7.1

NOTE: Some features are license-dependent so may not apply to user-specific installation of VIA. For example, CNV from NGS and Seq Var analysis capabilities are only available to those with a license that includes capabilities to process NGS data.

Aneusomy Workflow

1. Introduced a new step in **Processing** for users to analyze Aneusomy for Whole Chromosome or Whole chromosome and chromosomal arms using aberrant cell fraction.

2. A new **Aneusomy** tab in the **Sample Review** panel, displays the aneusomy call Gain/Loss, the estimated copy number, and a confidence value for the calls. Gains are displayed in blue text and losses are displayed in red text.

3. Chromosome number headers are hyperlinked to the specific region in the tracks i.e., the whole chromosome or the specific arm.

4. Copy Number estimates are displayed to 2 decimal places and the tooltip displays the underlying number.

5. Data derived from the P-arm of acrocentric chromosomes (Chromosomes: 13, 14, 15, 21 and 22) are excluded from the Whole Chromosome Aneusomy calculation.

6. A confidence score is included with each aneusomy call ranging from 0 to 1, representing our certainty in the existence (or non-existence) of the aneusomy.

7. Scrollbars allow access to the contents of the **Aneusomy** table.

8. The **Aneusomy** table is only present for Processing Types with aneusomy calling settings.

9. A tooltip in the **Aneusomy** panel explains the meaning of the confidence scores.

10. Aberrant cell fraction thresholds used as in the Aneusomy step are displayed in the sample information as well as in the **Aneusomy** table.

11. Resetting or duplicating a sample clears the aneusomy results.

12. Aneusomy calls persist between sessions and users.

13. A whole chromosome aneusomy call is not made when disparity is measured between the associated chromosome arms, wherein a dash "-" is displayed indicating the impact of a chromosome arm level event impacting the ability to measure whole chromosome aneusomy.

14. Processing settings already in use do not compute the Aneusomy call, they specify "None" by default for the Aneusomy step.

Enhancements to Circos Plot Visualization of Genomic Variants

EASILY INSPECT VARIANTS

1. Introduced the ability to zoom in and out on the Circos plot using the mouse wheel or trackpad or alternately zoom in to an area on the Circos plot using a left mouse click and drag.

2. Users can pan around the Circos plot view using the left mouse click to drag the view.

3. Users can reset the Circos plot view to the default view.

4. Left-clicking a variant on the Circos plot highlights it in the table.

5. Right-click a variant on the Circos plot to display a menu with options to view the variant in the tracks or view the variant details.

6. Improved tooltip on variants to present the list of genes (limited to 50) involved in a translocation in alphabetical order.

7. To provide a cleaner view the "Chr" text is not displayed anymore on the chromosome in the Circos plot.

8. Removed chrY from the Circos plot when a sample's gender is Female.

9. A new setting allows users to scale the Circos plot display for improved visualization in high DPI monitors.

10. A legend is available for the Circos plot.

CUSTOMIZED VIEW

1. Introduced preferences for Circos plot to display all chromosomes, only chromosomes with structural variants or a custom range of chromosomes.

2. Circos plot preferences allow the selection of Rainbow or Magenta color scheme for translocations.

3. Circos plot layout preferences can be persisted along with View Preferences.

4. The Circos plot and Whole Genome tab are displayed by default unless overridden by preferences.

IMPROVED VARIANT REPORTING

1. Users can annotate the Circos plot with gene names which are displayed in italicized font.

2. Gene annotations can be removed from the Circos plot.

3. Gene annotations added to the Circos plot persist between users and sessions.

4. The Circos plot along with the legend can be exported to the report using the <<I:Circos tab>> tag.

Reporting Improvements

New functionality has been introduced to automatically export all images and plots from the VIA UI into report templates by applying tags.

1. Introduced functionality to export images from the UI automatically into the report template:

   a. Insert the **Circos** plot into a report template using the tag <<I:Circos>>

   b. Insert the **Genomic Scars** plot into a report template using the tag <<I:Genomic Scars>>

   c. Insert the **Ideogram** plot into a report template using the tag «I:Ideogram»

   d. Insert the **Whole Genome** plot into a report template using the tag «I:Whole Genome»

2. The exported **Circos plot**, **Genomic Scars** plot and the **Ideogram** contain a legend with a key to the different events displayed by the visualization.

3. Reduced the number of significant figures for numerical data exported into a report template with values rounded up to 3 dps.

4. Removed the checkbox for **Remove CNV and AOH Events** so that it is no longer displayed in the copy to clipboard or export image of the **Genomic Scars** plot.

5. Improved user experience by greying out the down arrow icon in the **Sample Review** window to indicate that the sample type does not have an associated template.

6. Improved user experience by providing a checkbox to enable users to automatically open any report after saving.

General Improvements

1. OGM data visualization, classification, and reporting.

2. Updated the table so that it now displays the SMAP ID as **SV Event ID** in the event table for SVs derived from OGM data.

3. Implemented new functions for the DT to filter structural variants based on Length (SV_LENGTH), presence in the OGM control database (SV_PERCENTAGE_IN_OGM_CONTROLS) and quality (SV_QUALITY).

4. When a user applies a DT to classify only SV variants the name of the DT is displayed in the home page next to "Auto pre-classification."

5. Updated the display of OGM quality metrics in the sample info, table export and export to the Word report.

6. Truncated the display of the SV quality value displayed in the variant details panel to show only 2 dps.

7. Updated the OGM ISC nomenclature section to be more concise by displaying the genome build once and reordered events so that both CNVs and SVs derived from OGM are displayed within the same line. Events are organized in the following order: Sex-chromosome, Autosomes (in ascending order). Within each chromosome the CNV and allelic events are listed first followed by the SVs.

8. Added 3 new default attributes for OGM data for telomeric bias:

    a. Probability of High Telomeric Coverage Bias

    b. Probability of Moderate Telomeric Coverage Bias

    c. Prediction of Telomeric Coverage Bias

Sample Info

1. The phenotype selection in the sample info window includes **Clinical course** as a subset of **Clinical modifier**.

Sample Upload

1. Sample attribute values specified in the batch import descriptor file are now case insensitive.

Data Export

1. Expanded the JSON export schema so that it includes additional fields by default: %AOH, Estimated Gender, ISCN Nomenclature, Length, Genes, Gene count, Mosaic, Variant Interpretation, Cytoband and Event Significance (when the event is CNV from OGM BAM MSR).

2. Updated the JSON export so that it excludes data for unprocessed samples.

Similar Previous Cases

1. Updated the implementation of the Similar Previous cases for sequencing variants so that the data displayed and used for filtering corresponds to information about the region and the specific genotype change. In previous versions only the region information was used to generate these data.

2. Users can choose to include or exclude duplicated samples from similar previous case calculations.

3. Expanded DT functionality when using the DT SCORE (PREVIOUS_SIMILAR_CASES); users can now apply parameters to include or exclude duplicated samples from the count.

Sample Review

1. Disabled editing in the **ISCN** pop-up window in sample review.

2. Updated the text "**ISC nomenclature**" to "**ISCN**" throughout the software.

3. The **Events Table** is part of the **Display Layout** saved in the **Save View Preferences** menu.

4. A message is displayed when no UPD calls are detected by the UPD analysis.

5. The size filter accepts decimal values.

6. The gene track displays the gene orientation.

7. When a genotype is not available for a deletion the ISCN defaults to a heterozygous call (x1).

8. Improved the error message displayed when a user attempts to view sample information of a sample that has been deleted by another user.

9. To improve user experience the columns in the Guideline panel cannot be reordered.

10. Improved the export and copy to clipboard of the track view, so that it is exported as shown in the track even when the user has adjusted the scrollbar.

Home Page

1. The "selected" variant counts are displayed in the **Home** page after a sample has been locked.

2. The genome build of a sample is displayed on the **Home** page along with the "sample type," "data type" and "processing type."

3. Improved the error message displayed when a user attempts to add a report template to a sample type that does not contain the sample attributes specified in the report template.

4. A Client Version and Server Version are displayed in the sample information and added to the table export after a sample is locked.

5. Users can reset or reprocess samples when the sample is in an error state.

6. **Font size Preferences** includes font size of 16 that can be applied to the **Home** page, tracks and Circos plot.

Admin Tab

1. Improved the functionality and error messaging when adding duplicate labels to sample attributes.

2. Introduced functionality to prevent admin from adding redundant sample attributes and gene panels with whitespaces and workflow stages with different case.

3. Admin sample type tabs have been reordered so **Guidelines** are now displayed to the left of **Reports**.

4. Updated the text in the dropdown in the **Processing Types** section lists so that now **SNP FASST3** is displayed instead of **SNP-FASST3** (the dash has been deleted).

5. Updated the text displayed in the **Add User** dialogue to remove "Ability to" so that the texts for different privileges are shortened. For instance, "Ability to Perform admin operations" is now displayed as "Perform admin operations."

6. A new column has been added to the **Admins > Users** tab so that admins can now easily identify which users have admin permissions.

Methylation Samples

1. Improved the display of Illumina methylation samples by listing both idat files (red and green) in the sample info panel.

Bug Fixes in VIA Version 7.1

Changes to the Segmentation Algorithm

1. To address the behavior of copy number calls being made on chromosome Y for female samples, the probes on chromosome Y are being excluded as part of the pre-processing step for copy number segmentation for female samples. Removal of the nullizygous probes on chromosome Y from the probability calculation can also have a slight impact on segmentation for any chromosome, as all incoming probes are used to estimate probe LogR/BAF noise, which ultimately determines the strength of correlation between probe LogR/BAF and segmentation state by informing the shape of the probability distributions. This preprocessing step occurs for all CN algorithms in VIA 7.0. This change is not implemented for Agilent CGH and Custom CGH (non-SNP) data types. Internal testing has observed the impact to result in minor segmentation differences between software versions.

2. A bug was fixed where a CNV call was missed when some BAF probe values were exactly at 0.5. This has now been rectified to improve the accuracy of segmentation for the FASST2, SNP FASST2, FASST3 and SNP FASST3 algorithms.

3. To prevent segments from spanning across PAR and non-PAR regions in the case of Rank and SNP Rank segmentation algorithms, the PAR and non-PAR regions are segmented separately.

Other Bug Fixes

1. Resolved an issue which allowed Nirvana VCFs to be processed even when there was conflict between the genome build in the Sample Type and the file loaded. VIA 7.1 displays an error message **Position out of bounds** to indicate the discrepancy.

2. Rectified an issue that caused the **Open** button in the file/folder selection window to be greyed out when exporting results in JSON using VIA on Mac and Windows clients.

3. Addressed a bug that resulted in an **Access Denied** error message when importing Illumina final report samples.

4. Improved the error messaging displayed when CRAM files had missing header information. When the file is processed VIA displays the following error: "Error: FASTA reference file is not specified in the URI field of the CRAM header." When the file is used in the Multi Scale Reference Builder the error message reads "FASTA reference file is not specified in the URI field of the CRAM header."

5. Improved sample resetting so that ISCN, notes and sample interpretation are removed from the sample information once a sample is reset.

6. Resolved a thread race condition that can result in an error during SSL log in when "**use htpps"** is enabled.

7. Display a warning message to users when editing variant interpretation on the events that have been filtered out.

8. Resolved an issue which resulted in the SV events track showing an empty track for samples without SV data. In this update, if there is no SV data then the SV track is not displayed.

9. Addressed an issue which caused the **Similar Previous Cases** column to appear blank for an event that was being edited by one user while simultaneously being reviewed by another user.

10. Improved the display of the phenotypes ID column in the sample information which is no longer cut off and the whole phenotype ID is displayed; the columns are now resizable.

11. Resolved an issue that caused an error when users attempted to open a gene panel with a region with identical start and end coordinates.

12. Fixed an issue that caused an error message when a user changes the review status and immediately tries to close the sample.

13. Fixed the status message when Illumina data is batch imported and user selects to skip or overwrite.

14. Improved the scrolling bar in the OGM Molecules track to avoid the generation of errors.

15. Resolved a bug that caused an error message when a user removed all the tracks from the track display.

16. Renamed the default file name used when exporting all events of samples from the **Home** page to sample-event-export.json.gz.

17. Resolved an issue that prevented users from updating the processing unit after restarting the computer without shutting down the processing unit.

18. Resolved an error when the user attempts to logout when the server is down.

19. Now when the Admin adds "Explanation of DT" column to the Events Table, the decision tree results are displayed immediately after the spinning icons disappear.

20. Fixed a bug that allowed for the Illumina final report GSA assay type samples to be processed even when there was a discrepancy between genome builds.

21. Improved user experience by providing a warning when editing a note of an event that will not be visible after the filter process completes (this is applicable when users are editing notes while filtering is in progress).

22. Resolved an issue which caused conflicting parent of origin probe coloring for Gain and AOH events. With this update, the probe coloring on the BAF track will always match the informative probe counts for the AOH.

23. Fixed a bug that caused the report dropdown to display a blank white rectangle when it was disabled.

24. Fixed an issue where the table divider position failed to update when users switched back to the default view from a custom view.

Known issues Version 7.1

1. Duplicating or resetting a sample does not remove the gene label annotations from the Circos plot.

Technical Assistance

For technical assistance, contact Bionano Technical Support.

You can retrieve documentation on Bionano products, SDS's, certificates of analysis, frequently asked questions, and other related documents from the Support website or by request through e-mail and telephone.

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